Phenylketonuria (PKU)

Phenylketonuria is an inherited autosomal recessive disorder, which prevents the body from using the amino acid phenylalanine (Phe) properly. It is primarily a deficiency of the liver enzyme phenylalanine hydroxylase. Variant forms are caused by impaired synthesis or recycling of the biopterin (BH4) cofactor. Early detection and treatment is imperative to prevent mental retardation

Estimated Incidence (MI):	1:9,000 (includes Classic PKU, Mild PKU and non-PKU hyperphenylalaninemia)
Laboratory Screening Test:	Phenylalanine and Phenylalanine/Tyrosine ratio using Tandem Mass Spectrometry
Timing of Test:	≥ 24 hours of age: Results are valid
Feeding Effect:	Minimal, Tandem Mass Spectrometry can detect elevations in phenylalanine earlier than previously used qualitative methods.
Transfusion effect:	None
Confirmation:	All strong and persistent borderline positive tests are referred to the Pediatric Neurology Metabolic Clinic (PNMC) (734) 763-4697 **Do not send diagnostic labs before contacting PNMC.**
Treatment:	Phenylalanine free infant formula that should be started as soon after birth as possible **once the diagnosis has been confirmed.** Long-term treatment consists of maintaining a low phenylalanine diet for life through the use of special formula and low protein food products.

For further information, contact the Newborn Screening Program
Telephone (517) 335-9205
mdch-newbornscreening@michigan.gov

4/2003